https://doi.org/10.17576/JSA.2017.0701.15

Case Report

A Case of Gastrointestinal Stromal Tumour Mimicking Uterine Fibroid

Nurhayati A[1], Aida Hani MK (✉)[1], Nik Muhd Aslan A[2], Reena Rahayu MZ[3], Ani Amelia Z[1]

[1]Department of Obstetrics and Gynaecology, [2]Department of Radiotherapy & Oncology, [3]Department of Pathology, Faculty of Medicine, Universiti Kebangsaan Malaysia Medical Centre, Jalan Yaacob Latif, Bandar Tun Razak, 56000 Cheras, Kuala Lumpur, Malaysia.

Abstract

Gastrointestinal stromal tumour (GIST) is extremely rare with reported incidence of 20 per million per year. It is the most common mesenchymal tumour of the gastrointestinal tract. When it occurs at the pelvis in a female patient, it can be misleading to a gynaecological diagnosis. Non gynaecological diagnosis such as GIST must be considered in patients with pelvic mass presenting with atypical symptoms.

Keywords: Fibroid, gastrointestinal stromal tumour, mass, mesenchymal tumour, pelvic, rectal bleeding

Correspondence:

Aida Hani Mohd Kalok. Department of Obstetrics and Gynaecology, Faculty of Medicine, Universiti Kebangsaan Malaysia Medical Centre, Jalan Yaacob Latif, Bandar Tun Razak, 56000 Cheras, Kuala Lumpur, Malaysia. Tel: +603 91455950. Fax: +603 91456672 E-mail: aida.kalok@gmail.com

Date of submission: 9 Feb, 2017 Date of acceptance: 4 Apr, 2017

Introduction

Gastrointestinal stromal tumour (GIST) is a rare condition, with a reported incidence of 20 per million per year. It is the most common mesenchymal tumour of the gastrointestinal tract. Histologically, it ranges from pure spindle cell neoplasm to epithelioid neoplasms. Historically, GIST was categorized as smooth muscle tumours. Thus, smooth muscle tumours histologically comprising spindle cells, arising in the pelvis may easily be mistaken with a gynaecological disorder such as uterine fibroid or ovarian fibroma. We report a diagnostically challenging case of GIST mimicking uterine fibroid.

Case Report

A 49-year-old, nulliparous teacher, complained of lower abdominal pain, with lethargy and pelvic mass. She was diagnosed to have severe iron deficiency anaemia secondary to heavy menstrual bleeding due to uterine fibroid at another centre four months prior, and was planned for hysterectomy. She later developed per rectal bleeding with diarrhea. Abdominal examination revealed a soft but tender abdomen with a mass of 14 weeks size of gravid uterus. Full blood count showed mild leucocytosis, white cell count 13.4 x 10^9/l, haemoglobin 12.3g/dl and platelet 313 x 10^9/l. Tumor markers were not taken as the working diagnosis was uterine fibroid. Pelvic sonography noted an anteverted uterus with a 10.2 x 7.3cm solid cystic mass adherent to the anterior part of uterus (Fig. 1). Colonoscopy results were normal. CT scan findings noted that the mass was in continuity with the anterior part of uterus, suggestive of uterine fibroid.

Our impression was cystic degeneration of fibroid. We counseled for surgery as the pain was worsening, namely for total abdominal hysterectomy bilateral salpingooopherectomy (TAHBSO) as she was 49 years old with no fertility issue.

Abdominal incision was via Pfannenstiel incision. There was a friable mass adherent to a segment of small bowel and to the anterior uterus (Fig. 2). Small bowel resection with primary anastomosis was performed by the surgical team followed by TAHBSO. Specimens were sent for urgent histological examination (HPE).

https://doi.org/10.17576/JSA.2017.0701.15

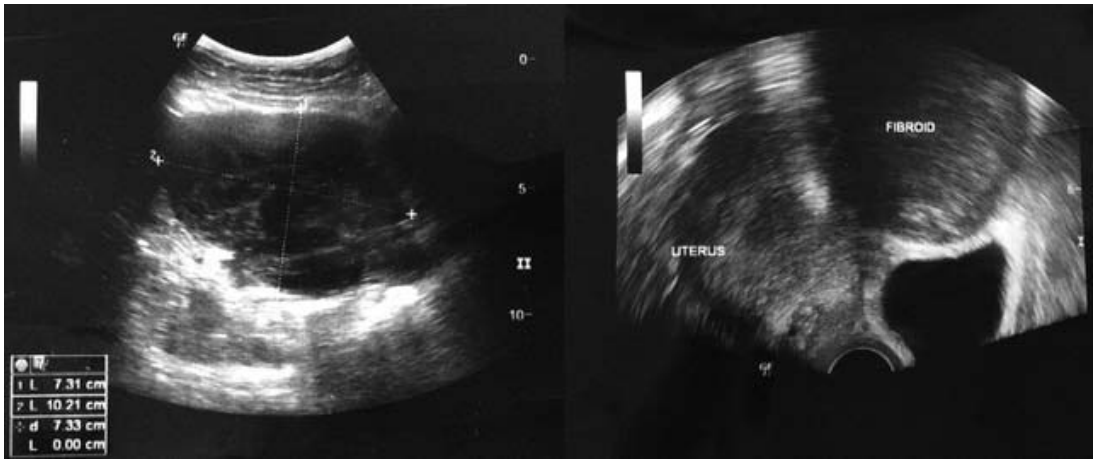

Figure 1: Transabdominal sonography (on the left) and transvaginal sonography (on the right) noted presence of a pelvic mass adherent to anterior uterus.

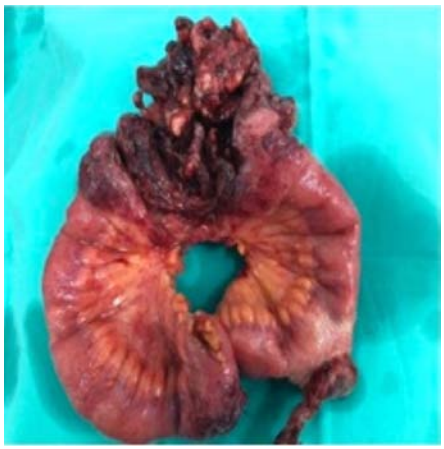

Figure 2: A cut segment of the small bowel with the tumour growing extraluminally.

Postoperative recovery was uncomplicated. HPE of the mass and the small bowel section showed malignant gastrointestinal tumour (GIST), high-risk category, whilst that of the uterus was leiomyomata with adenomyosis. Immunohistochemistry staining showed strong positivity for CD117 and DOG-1 with proliferative index for Ki-67 of 50%. Staining for CKAE1/AE3, smooth muscle actin and desmin were negative.

She is currently under the oncology team follow-up. She was counselled for imatinib therapy with six monthly CT scans for tumour surveillance.

Discussion

GIST is extremely rare, with a reported incidence of 20 per million per year, where 20-30% are malignant (1). It is the most common mesenchymal tumour of the gastrointestinal tract, the majority arising in the stomach. Even though GIST arises predominantly in the gastrointestinal tract, a small percentage (9%) arises extraintestinally (2).

Histologically, it ranges from pure spindle cell neoplasm to epithelioid neoplasms. Historically, GIST was categorized as smooth muscle tumours. Thus, smooth muscle tumours histologically comprising spindle cells, arising in the pelvis may easily be mistaken with a gynaecological disorder such as uterine leiomyoma or ovarian fibroma (3). However, with the advent of molecular therapies targeting CD117; also known as KIT proto-oncogene; GIST must be distinguished from smooth muscle tumours. Approximately, 90% of GIST tumors are strongly positive for KIT (CD117). A small percentage of GIST is KIT negative but is strongly positive for DOG1, a recently developed immunohistochemical marker which is relatively sensitive and specific for GIST.

In this case, as the mass was adherent to the uterus, and it grew extraluminally from the small bowel, it created a diversion from the actual diagnosis.

Literature review using PubMed have reported similar diagnostic dilemma. Two cases were reported by Morimura et al. (3), where patients were operated initially for suspected ovarian theco-fibroma and subserosal fibroma, however intraoperative findings and HPE confirmed GIST. Another author (4) also reported a case of suspected ovarian carcinoma, in a patient presenting with a large abdominal mass which on CT scan appeared to be multiseptated with ascites, turned out to be GIST.

There is no recommendation at present regarding the best imaging modality to diagnose GIST. Most of the diagnoses were made either from intra-operatively or during histological examination. However, current

https://doi.org/10.17576/JSA.2017.0701.15

guidelines advocate CT scans as part of the initial evaluation and staging to detect distant metastases. MRI may also be useful in evaluation of rectal GIST for resectability (5).

Initially, the mainstay treatment for localized GIST was complete surgical resection. However recently, Imatinib, an oral tyrosine kinase inhibitor, was introduced as an adjuvant therapy. Studies have shown a survival benefit of three years in high-risk patients; 5 year survival rate of 92% with 3 years therapy versus 81.7% with 1 year therapy (6,7). Currently, high-risk patients with risk of relapse should be offered adjuvant Imatinib as standard treatment (5). Unfortunately, it may be too costly for some as in this case.

Conclusion

The rarity of GIST and the highly variable clinical presentations and appearances on radiological imaging, impose challenges in making the diagnosis. Gastrointestinal symptoms in a female patient with a pelvic mass must raise the suspicion of possible non-gynaecological pathology, and differential diagnoses should include GIST.

References

1. Stamatakos M, Douzinas E, Stefanaki C, et al. Review- Gastrointestinal stromal tumor. World J Surg Oncol 2009; 7: 61.

2. DeMatteo RP, Lewis JJ, Leung D, Mudan SS, Woodruff JM, Brennan MF. Two hundred gastrointestinal stromal tumors: recurrence patterns and prognostic factors for survival. Ann Surg 2000; 231(1): 51-8.

3. Morimura Y, Yamashita N, Koyama N, et al. Gastrointestinal stromal tumor mimicking gynecological disease. Fukushima J Med Sci 2006; 52 (l): 21-8.

4. Lee T. Gastrointestinal stromal tumor (GIST) presenting as a huge pelvic mass. Geburtshilfe Frauenheilkd 2013; 73(1): 70–3.

5. ESMO/European Sarcoma Network Working Group. Gastrointestinal stromal tumours: ESMO Clinical Practice Guidelines for for diagnosis,treatment and follow-up. Ann Oncol 2014; 25 Suppl 3: iii21-6.

6. Joensuu H, Eriksson M, Sundby Hall K, et al. One vs three years adjuvant imatinib for operable gastrointestinal stromal tumor: a randomized trial. JAMA 2012; 307(12): 1265-72.

7. DeMatteo RP, Ballman KV, Antonescu CR, et al. Adjuvant imatinib mesylate after resection of localised, primary gastrointestinal stromal tumour: a randomised, double-blind, placebo-controlled trial. Lancet 2009; 373(9669): 1097-104.